



LET'S TALK GENETICS

Screening and Testing for SLO

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Genetic Counselor

DHCR7 Carrier Frequencies

- 2017 study of 262,399 people
- No family history
- Purpose was routine carrier screening

1 in 40

CARRIER RATE

Most common in individuals with Northern European ancestry,
but seen in all ethnic groups .

DHCR7 Carrier Frequencies

**AFRICAN
AMERICANS**

1 in 183

**ASHKENAZI
JEWISH**

1 in 43

**NORTHERN
EUROPEANS**

1 in 54

HISPANIC

1 in 167

**SOUTHERN
EUROPEAN**

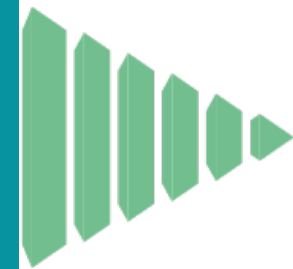
1 in 66

**MIXED/ OTHER
CAUCASIAN**

1 in 55

DHCR7 gene

CODES FOR ENZYME

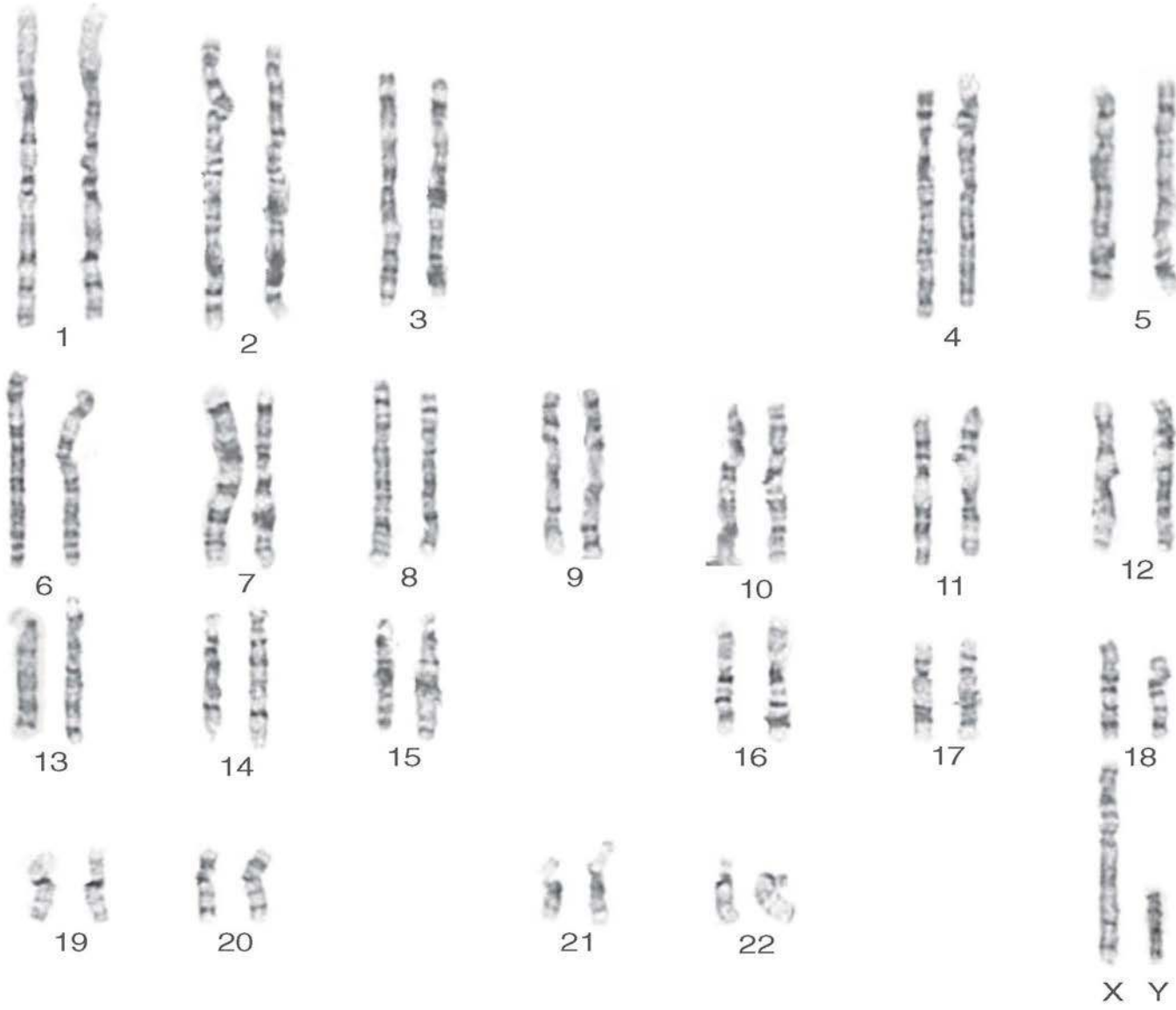


7-DHC reductase

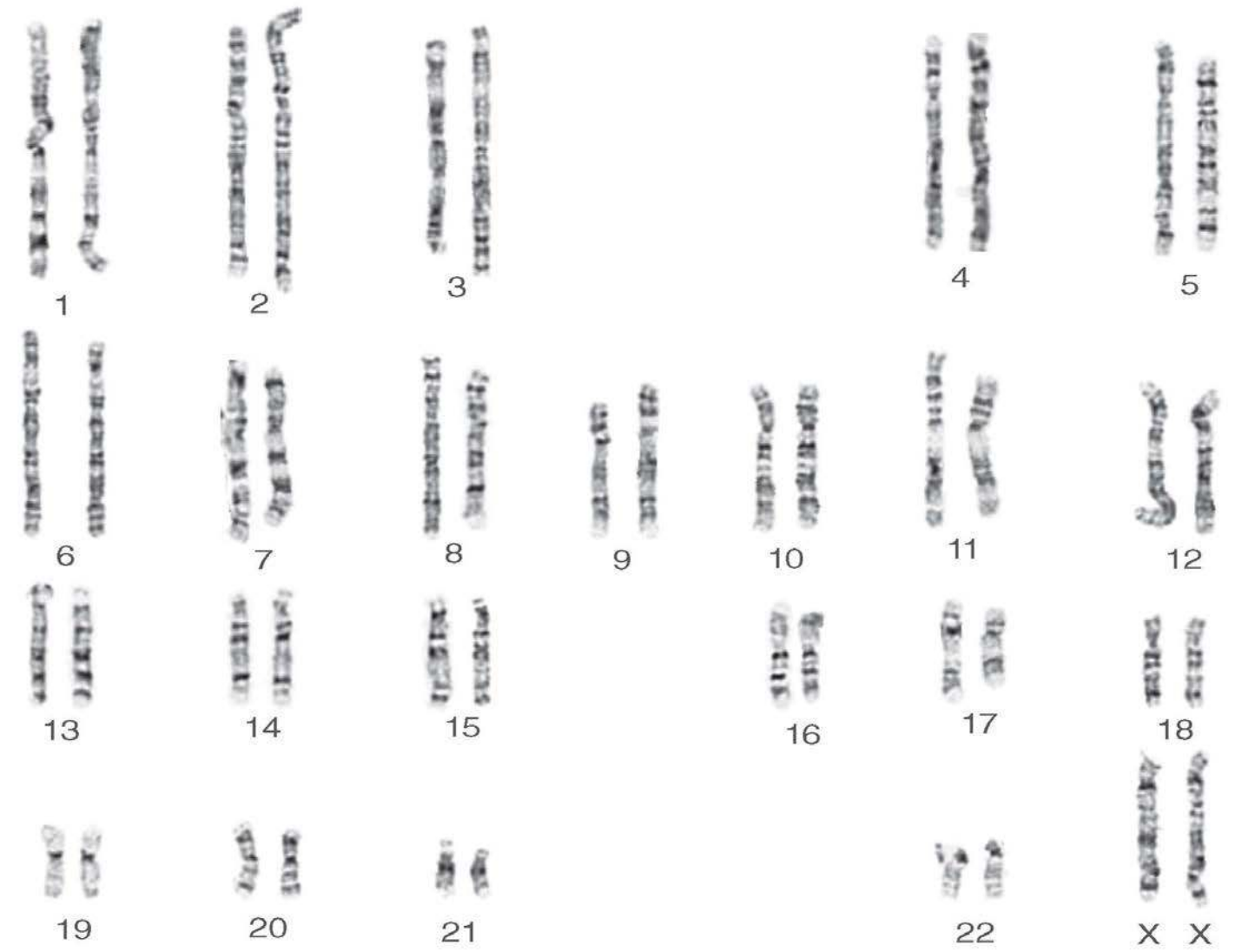
ENZYME MAKES
CHOLESTEROL



46, XY

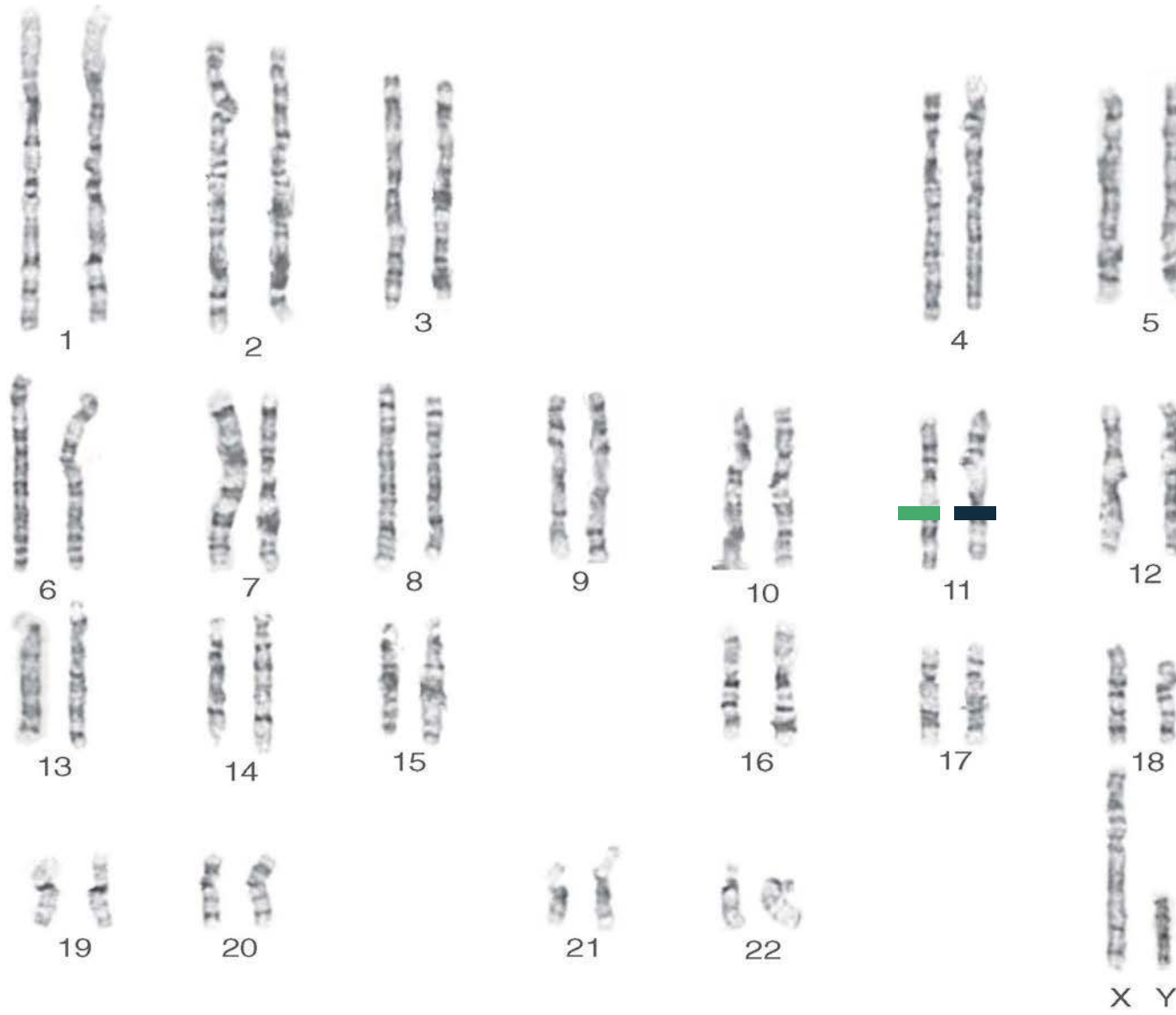


46, XX





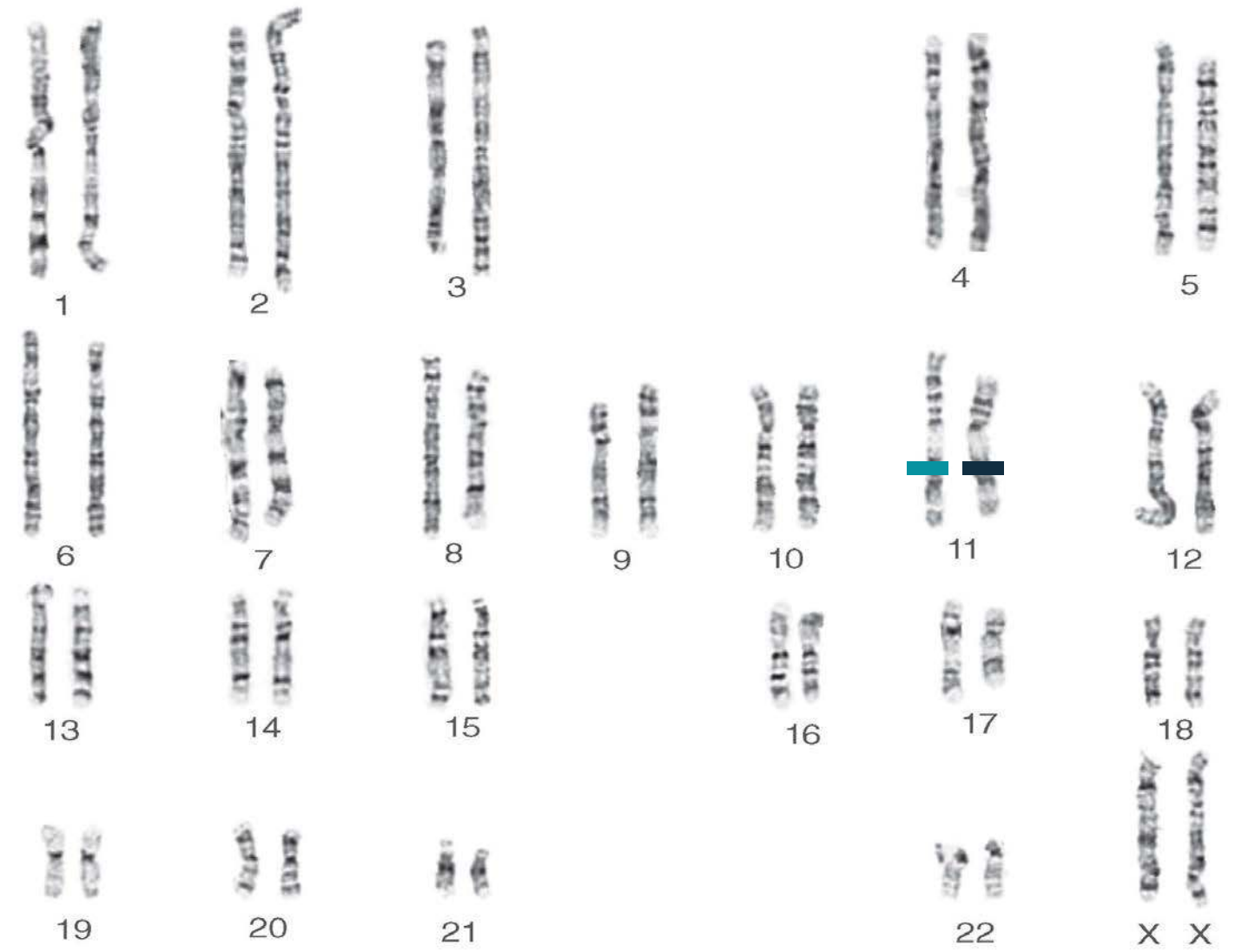
46, XY



Recessive inheritance = 1 in 4 chance



46, XX



What is a genetic variant?

c.964-1G>C

IVS8-1G>C

c.452G>A

W151*

c.278C>T

T93M



Testing for SLO syndrome

HOW IT WORKS

Standard Prenatal Testing and Screening Options

CVS

AMNIOCENTESIS

QUAD SC REEN

ULTRASOUND

Standard Prenatal Testing and Screening Options

CVS

- Diagnostic
- Sterol profile /gene
- Performed 10-12 weeks in pregnancy

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QUAD SCREEN

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- Diagnostic
- Sterol profile/gene
- After 14 weeks

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ULTRASOUND

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QUAD SCREEN

- Maternal serum at 16 weeks in pregnancy
- Measures markers
- Requires follow-up

ULTRASOUND

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QUAD SCREEN

- Maternal serum at 16 weeks in pregnancy
- Measures markers
- Requires follow-up

ULTRASOUND

- Not diagnostic
- Not specific
- Requires follow-up

MATERNAL URINE

- Diagnostic (validated) and non-invasive
- 14-22 weeks gestation
- Currently available in the UK
- Sample logistics

**Institute of Metabolism
and Systems Research
(IMSR) in Birmingham,
England**

PREIMPLANTATION GENETIC TESTING (PGT)

Newer Tests

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and Systems Research
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PREIMPLANTATION GENETIC TESTING (PGT)

- Diagnostic*
- Cost
- Requires IVF
- Specialized Centers

www.sharinghealthygenes.com

Newer Tests

Carrier Screening

AVAILABLE NOW

- Hundreds of hereditary conditions, \$250
- Do *before* pregnancy
- Expanded gene panel includes SLO gene
- Not all tests are equal



